



## GARS gene

glycyl-tRNA synthetase

### Normal Function

The GARS gene provides instructions for making an enzyme called glycyl-tRNA synthetase. This enzyme is found in all cell types and plays an important role in the production (synthesis) of proteins. During protein synthesis, building blocks (amino acids) are connected together in a specific order, creating a chain of amino acids. Glycyl-tRNA synthetase plays a role in adding the amino acid glycine at the proper place in a protein's chain of amino acids.

### Health Conditions Related to Genetic Changes

#### Charcot-Marie-Tooth disease

Researchers have identified a few mutations in the GARS gene that cause a form of Charcot-Marie-Tooth disease known as type 2D. These mutations change single amino acids used to make glycyl-tRNA synthetase. It is unclear how GARS gene mutations lead to type 2D Charcot-Marie-Tooth disease. The mutations probably reduce the activity of glycyl-tRNA synthetase. Scientists suggest that nerve cells may be especially sensitive to a decrease in the activity of this enzyme. In particular, a reduction in glycyl-tRNA synthetase activity may impair the ability of specialized outgrowths from nerve cells (axons) to transmit nerve impulses.

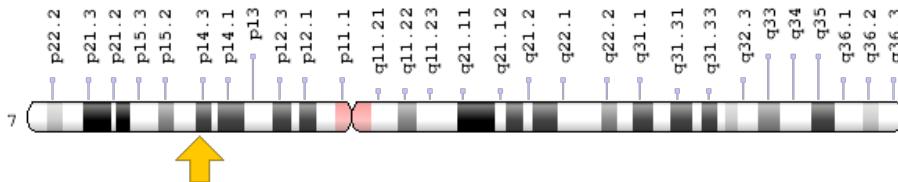
#### distal hereditary motor neuropathy, type V

Several GARS gene mutations have been identified in individuals with distal hereditary motor neuropathy, type V. These mutations change single amino acids used to make glycyl-tRNA synthetase. It is unclear how GARS gene mutations lead to this disorder. The mutations probably reduce the activity of glycyl-tRNA synthetase. As in Charcot-Marie-Tooth disease, a reduction in glycyl-tRNA synthetase activity may impair transmission of nerve impulses.

## Chromosomal Location

Cytogenetic Location: 7p14.3, which is the short (p) arm of chromosome 7 at position 14.3

Molecular Location: base pairs 30,594,735 to 30,634,033 on chromosome 7 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- CMT2D
- DSMAV
- glycine tRNA ligase
- GlyRS
- SMAD1
- SYG\_HUMAN

## Additional Information & Resources

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Specific Enzymes Couple Each Amino Acid to Its Appropriate tRNA Molecule  
<https://www.ncbi.nlm.nih.gov/books/NBK26829/#A1062>

### GeneReviews

- GARS-Associated Axonal Neuropathy  
<https://www.ncbi.nlm.nih.gov/books/NBK1242>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GARS%5BTIAB%5D%29+OR+%28glycyl-tRNA+synthetase%5BTIAB%5D%29+NOT+%28chromosome+21%5BTIAB%5D%29%29+OR+%28%28CMT2D%5BTIAB%5D%29+OR+%28glycyl+tRNA+synthetase%29+NOT+%2812q24%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

## OMIM

- GLYCYL-tRNA SYNTHETASE  
<http://omim.org/entry/600287>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_GARS.html](http://atlasgeneticsoncology.org/Genes/GC_GARS.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=GARS%5Bgene%5D>
- HGNC Gene Family: Aminoacyl tRNA synthetases, Class II  
<http://www.genenames.org/cgi-bin/genefamilies/set/132>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=4162](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4162)
- Inherited Peripheral Neuropathies Mutation Database  
<http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=27>
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/2617>
- UniProt  
<http://www.uniprot.org/uniprot/P41250>

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